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# Pxe.st25 SEQUENCE LISTING

<110> PXE International, Inc.
 University of Hawaii

<120> Mutations in a gene encoding an ABC transporter (MRP6) causing Pseudoxanthoma Elasticum

<130> PXE-001PC

<160> 27

<170> PatentIn version 3.0

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<213> Homo sapiens

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<221> misc\_feature

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Page 114

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Gly Thr Pro Glu Ala Pro Glu Phe Leu Ile His Pro Thr Val Trp Leu
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Gln Val Phe His Ser Thr Phe Leu Leu Gly Thr Leu Ser Leu Ile Ile 305 310 315 320

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Asn Ser Lys Thr Ile Lys Phe His Gly Trp Glu Gly Ala Phe Leu Asp 500 505 510

Met Arg Gln Lys Asp Ser Arg Ala Arg Leu Thr Ser Ser Ile Leu Arg

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Ile Glu Gly Ala Val Ala Tyr Val Pro Gln Glu Ala Trp Val Gln Asn

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TIP	ПСи	Olu	Arg	725	Бей	Giu	AIa	СуЗ	730	пец	GIII	rio	nsp	735	ASP
Ser	Phe	Pro	Glu 740	Gly	Ile	His	Thr	Ser 745	Ile	Gly	Glu	Gln	Gly 750	Met	Asn
Leu	Ser	Gly 755	Gly	Gln	Lys	Gln	Arg 760	Leu	Ser	Leu	Ala	Arg 765	Ala	Val	Tyr
Arg	Lys 770	Ala	Ala	Val	Tyr	Leu 775	Leu	Asp	Asp	Pro	Leu 780	Ala	Ala	Leu	Asp
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Cys	Leu 850	Leu	Asp	Gln	Ala	Arg 855	Gln	Pro	Gly	Asp	Arg 860	Gly	Glu	Gly	Glu
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- Pro Asp Arg Ala Gly Trp Pro Ala Gly Lys Asp Ser Ile Gln Tyr Gly 915 920 925
- Arg Val Lys Ala Thr Val His Leu Ala Tyr Leu Arg Ala Val Gly Thr 930 935 940
- Pro Leu Cys Leu Tyr Ala Leu Phe Leu Phe Leu Cys Gln Gln Val Ala 945 950 955 960
- Ser Phe Cys Arg Gly Tyr Trp Leu Ser Leu Trp Ala Asp Asp Pro Ala 965 970 975
- Val Gly Gln Gln Thr Gln Ala Ala Leu Arg Gly Gly Ile Phe Gly 980 985 990
- Leu Leu Gly Cys Leu Gln Ala Ile Gly Leu Phe Ala Ser Met Ala Ala 995 1000 1005
- Val Leu Leu Gly Gly Ala Arg Ala Ser Arg Leu Leu Phe Gln Arg 1010 1015 1020
- Leu Leu Trp Asp Val Val Arg Ser Pro Ile Ser Phe Phe Glu Arg 1025 1030 1035
- Thr Pro Ile Gly His Leu Leu Asn Arg Phe Ser Lys Glu Thr Asp 1040 1045 1050
- Thr Val Asp Val Asp Ile Pro Asp Lys Leu Arg Ser Leu Leu Met 1055 1060 1065
- Tyr Ala Phe Gly Leu Leu Glu Val Ser Leu Val Val Ala Val Ala 1070 1080

#### Pxe.st25 Thr Pro Leu Ala Thr Val Ala Ile Leu Pro Leu Phe Leu Leu Tyr Ala Gly Phe Gln Ser Leu Tyr Val Val Ser Ser Cys Gln Leu Arg Arg Leu Glu Ser Ala Ser Tyr Ser Ser Val Cys Ser His Met Ala Glu Thr Phe Gln Gly Ser Thr Val Val Arg Ala Phe Arg Thr Gln Ala Pro Phe Val Ala Gln Asn Asn Ala Arg Val Asp Glu Ser Gln Arg Ile Ser Phe Pro Arg Leu Val Ala Asp Arg Trp Leu Ala Ala Asn Val Glu Leu Leu Gly Asn Gly Leu Val Phe Ala Ala Ala Thr Cys Ala Val Leu Ser Lys Ala His Leu Ser Ala Gly Leu Val Gly Phe Ser Val Ser Ala Ala Leu Gln Val Thr Gln Thr Leu Gln Trp Val Val Arg Asn Trp Thr Asp Leu Glu Asn Ser Ile Val Ser Val Glu Arg Met Gln Asp Tyr Ala Trp Thr Pro Lys Glu Ala Pro Trp Arg Leu Pro Thr Cys Ala Ala Gln Pro Pro Trp Pro Gln Gly Gly

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Pxe.st25
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1

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49

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Leu Ser Leu Cys Phe Val Arg Ala Ala Ser Ser Trp Val Pro Pro Met 35 40 45
Tyr Leu Trp Val Leu Gly Pro Ile Tyr Leu Leu Tyr Ile His Arg His 50 55 60
Gly Arg Cys Tyr Leu Arg Met Ser His Leu Phe Lys Thr Lys Met Val 65 70 75 80
Leu Gly Leu Ala Leu Ile Leu Leu Tyr Thr Phe Asn Val Ala Val Pro 85 90 95
Leu Trp Arg Ile His Gln Gly Val Pro Gln Ala Pro Glu Leu Leu Ile 100 105 110
His Pro Thr Val Trp Leu Thr Thr Met Ser Phe Ala Thr Phe Leu Ile 115 120 125

# Pxe.st25 His Met Glu Arg Arg Lys Gly Val Arg Ser Ser Gly Val Leu Phe Gly Tyr Trp Leu Leu Cys Cys Ile Leu Pro Gly Ile Asn Thr Val Gln Gln Ala Ser Ala Gly Asn Leu Arg Gln Glu Pro Leu His His Leu Ala Thr Tyr Leu Cys Leu Ser Leu Val Val Ala Glu Leu Val Leu Ser Cys Leu Val Asp Gln Pro Pro Phe Phe Ser Glu Asp Ser Gln Pro Leu Asn Pro Cys Pro Glu Ala Glu Ala Ser Phe Pro Ser Lys Ala Met Phe Trp Trp Ala Ser Gly Leu Leu Trp Arg Gly Tyr Lys Lys Leu Leu Gly Pro Lys Asp Leu Trp Ser Leu Gly Arg Glu Asn Ser Ser Glu Glu Leu Val Ser Gln Leu Glu Arg Glu Trp Arg Arg Ser Cys Asn Gly Leu Pro Gly His Lys Gly His Ser Ser Val Gly Ala Pro Glu Thr Glu Ala Phe Leu Gln Pro Glu Arg Ser Gln Arg Gly Pro Leu Leu Arg Ala Ile Trp'Arg Val

Phe Arg Ser Thr Phe Leu Leu Gly Thr Leu Ser Leu Val Ile Ser Asp 305 310 315 320

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Met	Gly	Asp	Arg 340	Asn	Ser	Ser	Ala	Trp 345	Thr	Gly	Trp	Leu	Leu 350	Ala	Val
Leu	Met	Phe 355	Ala	Ala	Ala	Cys	Leu 360	Gln	Thr	Leu	Phe	Glu 365	Gln	Gln	His
Met	Tyr 370	Arg	Ala	Lys	Val	Leu 375	Gln	Met	Arg	Leu	Arg 380	Thr	Ala	Ile	Thr
Gly 385	Leu	Val	Tyr	Arg	Lys 390	Val	Leu	Val	Leu	Ser 395	Ser	Gly	Ser	Arg	Lys 400
Ser	Ser	Ala	Ala	Gly 405	Asp	Val	Val	Asn	Leu 410	Val	Ser	Val	Asp	Ile 415	Gln
Arg	Leu	Ala	Glu 420	Ser	Ile	Ile	Tyr	Leu 425	Asn	Gly	Leu	Trp	Leu 430	Leu	Phe
Leu	Trp	Ile 435	Phe	Val	Cys	Phe	Val 440	Tyr	Leu	Trp	Gln	Leu 445	Leu	Gly	Pro
Ser	Ala 450		Thr				Val						Pro	Leu	Asn
Phe 465	Phe	Ile	Thr	Lys	Lys 470	Arg	Gly	Phe	His	Gln 475	Glu	Glu	Gln	Met	Arg 480
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Arg	Thr	Ile	Lys 500	Ser	His	Gly	Trp	Glu 505	His	Ala	Phe	Leu	Glu 510	Arg	Leu

# Leu His Ile Arg Gly Gln Glu Leu Ser Ala Leu Lys Thr Ser Thr Leu 515 Leu Phe Ser Val Ser Leu Val Ser Phe Gln Val Ser Thr Phe Leu Val 530

- Ala Leu Val Val Phe Ala Val His Thr Leu Val Ala Glu Asp Asn Ala 545 550 560
- Met Asp Ala Glu Lys Ala Phe Val Thr Leu Thr Val Leu Ser Ile Leu 565 570 575
- Asn Lys Ala Gln Ala Phe Leu Pro Phe Ser Val His Cys Ile Val Gln 580 585 590
- Ala Arg Val Ser Phe Asp Arg Leu Ala Ala Phe Leu Cys Leu Glu Glu 595 600 605
- Val Asp Pro Asn Gly Met Ile Ala Ser Asn Ser Arg Arg Ser Ser Lys 610 620
- Asp Arg Ile Ser Val His Asn Gly Thr Phe Ala Trp Ser Gln Glu Ser 625 630 635 640
- Pro Pro Cys Leu His Gly Ile Asn Leu Thr Val Pro Gln Gly Cys Leu 645 650 655
- Leu Ala Val Val Gly Pro Val Gly Ala Gly Lys Ser Ser Leu Leu Ser 660 665 670
- Ala Leu Leu Gly Glu Leu Leu Lys Val Glu Gly Ser Val Ser Ile Glu 675 680 685
- Gly Ser Val Ala Tyr Val Pro Gln Glu Ala Trp Val Gln Asn Thr Ser 690 695 700

								Pxe.s							
Val 705	Ala	Glu	Asn	Val	Cys 710	Phe	Arg	Gln	Glu	Leu 715	Asp	Leu	Pro	Trp	Leu 720
Gln	Lys	Val	Leu	Asp 725	Ala	Cys	Ala	Leu	Gly 730	Ser	Asp	Val	Ala	Ser 735	Phe
Pro	Ala	Gly	Val 740	His	Thr	Pro	Ile	Gly 745	Glu	Gln	Gly	Met	Asn 750	Leu	Ser
Gly	Gly	Gln 755	Lys	Gln	Arg	Leu	Ser 760	Leu	Ala	Arg	Ala	Val 765	Tyr	Lys	Lys
Ala	Ala 770	Ile	Tyr	Leu	Leu	Asp 775	Asp	Pro	Leu	Ala	Ala 780	Leu	Asp	Ala	His
Val 785	Ser	Gln	Gln	Val	Phe 790	Lys	Gln	Val	Ile	Gly 795	Pro	Ser	Gly	Leu	Leu 800
Gln	Gly	Thr	Thr	Arg 805	Ile	Leu	Val	Thr	His 810	Thr	Leu	His	Val	Leu 815	Pro
Gln	Ala	Asp	Arg 820	Ile	Leu	Val	Leu	Ala 825	Asn	Gly	Thr	Ile	Ala 830	Glu	Met
Gly	Ser	Tyr 835	Gln	Asp	Leu	Leu	Gln 840	Arg	Asn	Gly	Ala	Leu 845	Val	Gly	Leu
Leu	Asp 850	Gly	Ala	Arg	Gln	Pro 855	Ala	Gly	Thr	His	Asp 860	Ala	Ala	Thr	Ser
Asp 865	Asp	Leu	Gly	Gly	Phe 870	Pro	Gly	Gly	Gly	Arg 875	Pro	Thr	Cys	Arg	Pro 880

890

895

Asp Arg Pro Arg Pro Thr Glu Ala Ala Pro Val Lys Gly Arg Ser Thr

- Ser Glu Val Gln Met Glu Ala Ser Leu Asp Asp Pro Glu Ala Thr Gly 900 905 910
- Leu Thr Ala Glu Glu Asp Ser Val Arg Tyr Gly Arg Val Lys Ile Thr 915 920 925
- Ile Tyr Leu Ser Tyr Leu Arg Ala Val Gly Thr Pro Leu Cys Thr Tyr 930 940
- Thr Leu Phe Leu Cys Gln Gln Val Ala Ser Phe Ser Gln Gly 945 950 955 960
- Tyr Trp Leu Ser Leu Trp Ala Asp Asp Pro Val Val Asp Gly Arg Gln 965 970 975
- Met His Ala Leu Arg Gly Trp Val Phe Gly Leu Leu Gly Cys Leu 980 985 990
- Gln Ala Ile Gly Leu Phe Ala Ser Met Ala Ala Val Phe Leu Gly Gly 995 1000 1005
- Ala Arg Ala Ser Gly Leu Leu Phe Arg Ser Leu Leu Trp Asp Val 1010 1015 1020
- Ala Arg Ser Pro Ile Gly Phe Phe Glu Arg Thr Pro Val Gly Asn 1025 1030 1035
- Leu Leu Asn Arg Phe Ser Lys Glu Thr Asp Thr Val Asp Val Asp 1040 1045 1050
- Ile Pro Asp Lys Leu Arg Ser Leu Leu Thr Tyr Ala Phe Gly Leu 1055 1060 1065
- Leu Glu Val Gly Leu Ala Val Thr Met Ala Thr Pro Leu Ala Ile 1070 1075 1080

## Pxe.st25 Val Ala Ile Leu Pro Leu Met Val Leu Tyr Ala Gly Phe Gln Ser Leu Tyr Val Ala Thr Ser Cys Gln Leu Arg Arg Leu Glu Ser Ala Arg Tyr Ser Ser Val Cys Ser His Met Ala Glu Thr Phe Gln Gly Ser Leu Val Val Arg Ala Phe Arg Ala Gln Ala Ser Phe Thr Ala Gln His Asp Ala Leu Met Asp Glu Asn Gln Arg Val Ser Phe Pro Lys Leu Val Ala Asp Arg Trp Leu Ala Thr Asn Leu Glu Leu Leu Gly Asn Gly Leu Val Phe Val Ala Ala Thr Cys Ala Val Leu Ser Lys Ala His Leu Ser Ala Gly Leu Val Gly Phe Ser Val Ser Ala Ala Leu Gln Val Thr Gln Thr Leu Gln Trp Val Val Arg Ser Trp Thr Asp Leu Glu Asn Ser Met Val Ala Val Glu Arg Val Gln Asp Tyr Ala Arg Ile Pro Lys Glu Ala Pro Trp Arg Leu Pro Thr Cys Ala Ala Gln Pro Leu Trp Pro Cys Gly Gly Gln Ile Glu Phe Arg

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## Pxe.st25 Asp Phe Gly Leu Arg His Arg Pro Glu Leu Pro Leu Ala Val Gln Gly Val Ser Leu Lys Ile His Ala Gly Glu Lys Val Gly Ile Val Gly Arg Thr Gly Ala Gly Lys Ser Ser Leu Ala Trp Gly Leu Leu Arg Leu Gln Glu Ala Ala Glu Gly Asn Ile Trp Ile Asp Gly Val Pro Ile Thr His Val Gly Leu His Thr Leu Arg Ser Arg Ile Thr Ile Ile Pro Gln Asp Pro Val Leu Phe Pro Gly Ser Leu Arg Met Asn Leu Asp Leu Leu Gln Glu His Thr Asp Glu Gly Ile Trp Ala Ala Leu Glu Thr Val Gln Leu Lys Ala Phe Val Thr Ser Leu Pro Gly Gln Leu Gln Tyr Glu Cys Ala Gly Gln Gly Asp Asp Leu Ser Val Gly His Lys Gln Leu Leu Cys Leu Ala Arg Ala Leu Leu Arg Lys Thr Gln Ile Leu Ile Leu Asp Glu Ala Thr Ala Ser Val Asp Pro Gly Thr Glu Met Gln Met Gln Ala Ala Leu Glu Arg Trp Phe

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                         1450
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Met Asp Cys Ala Arg Val Leu Val Met Asp Glu Gly Gln Val Ala
    1460
                         1465
                                              1470
Glu Ser Gly Asn Pro Ala Gln Leu Leu Ala Gln Lys Gly Leu Phe
    1475
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                                              1485
Tyr Arg Leu Ala His Glu Ser Gly Leu Ala
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